



Townes-Brocks Syndrome

Townes-Brocks syndrome is a genetic condition that affects several parts of the body. The most common features of this condition are an obstruction of the anal opening (imperforate anus), abnormally shaped ears, and hand malformations that most often affect the thumb. Most people with this condition have at least two of these three major features.

Other possible signs and symptoms of Townes-Brocks syndrome include kidney abnormalities, mild to profound hearing loss, heart defects, and genital malformations. These features vary among affected individuals, even within the same family. Intellectual disability or learning problems have also been reported in about 10 percent of people with Townes-Brocks syndrome.

Frequency

The prevalence of this condition is unknown, although one study estimated that it may affect 1 in 250,000 people. It is difficult to determine how frequently Townes-Brocks syndrome occurs because the varied signs and symptoms of this disorder overlap with those of other genetic syndromes.

Genetic Changes

Mutations in the *SALL1* gene cause Townes-Brocks Syndrome. The *SALL1* gene is part of a group of genes called the SALL family. These genes provide instructions for making proteins that are involved in the formation of tissues and organs before birth. SALL proteins act as transcription factors, which means they attach (bind) to specific regions of DNA and help control the activity of particular genes. Some mutations in the *SALL1* gene lead to the production of an abnormally short version of the SALL1 protein that malfunctions within the cell. Other mutations prevent one copy of the gene in each cell from making any protein. It is unclear how these genetic changes disrupt normal development and cause the birth defects associated with Townes-Brocks syndrome.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- anal-ear-renal-radial malformation syndrome
- deafness-imperforate anus-hypoplastic thumbs syndrome

- imperforate anus-hand and foot anomalies syndrome
- renal-ear-anal-radial syndrome (REAR)
- sensorineural deafness-imperforate anus-hypoplastic thumbs syndrome
- Townes syndrome

Diagnosis & Management

These resources address the diagnosis or management of Townes-Brocks Syndrome:

- GeneReview: Townes-Brocks Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1445>
- Genetic Testing Registry: Townes syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265246/>
- MedlinePlus Encyclopedia: Ear Disorders (image)
<https://medlineplus.gov/ency/imagepages/17281.htm>
- MedlinePlus Encyclopedia: Imperforate Anus
<https://medlineplus.gov/ency/article/001147.htm>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Ear Disorders (image)
<https://medlineplus.gov/ency/imagepages/17281.htm>
- Encyclopedia: Imperforate Anus
<https://medlineplus.gov/ency/article/001147.htm>
- Health Topic: Hand Injuries and Disorders
<https://medlineplus.gov/handinjuriesanddisorders.html>

- Health Topic: Hearing Disorders and Deafness
<https://medlineplus.gov/hearingdisordersanddeafness.html>
- Health Topic: Kidney Diseases
<https://medlineplus.gov/kidneydiseases.html>

Genetic and Rare Diseases Information Center

- Townes-Brocks syndrome
<https://rarediseases.info.nih.gov/diseases/7784/townes-brocks-syndrome>

Educational Resources

- American Society for Surgery of the Hand: Congenital Hand Differences
<http://www.assh.org/handcare/hand-arm-conditions/Congenital-Differences>
- Cincinnati Children's Hospital Medical Center: Anorectal Malformations
<https://www.cincinnatichildrens.org/health/a/anorectal-malformations>
- Disease InfoSearch: Townes-Brocks syndrome
<http://www.diseaseinfosearch.org/Townes-Brocks+syndrome/7145>
- MalaCards: townes-brocks syndrome
http://www.malacards.org/card/townes_brocks_syndrome
- Orphanet: Townes-Brocks syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=857

Patient Support and Advocacy Resources

- National Association of the Deaf
<https://www.nad.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/townes-brocks-syndrome/>
- Resource list from the University of Kansas Medical Center: Deafness and Hard of Hearing
<http://www.kumc.edu/gec/support/hearing.html>

GeneReviews

- Townes-Brocks Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1445>

Genetic Testing Registry

- Townes syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265246/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28townes-brocks+syndrome%5BTIAB%5D%29+OR+%28townes+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- TOWNES-BROCKS SYNDROME
<http://omim.org/entry/107480>

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<https://ghr.nlm.nih.gov/condition/townes-brocks-syndrome>

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